American Journal of Medical Genetics 69:221 (1997)

Letter to the Editor

Circumferential Ringed Creases ("Michelin Tire Babies") With Specific Histologic Findings and/or Karyotype Abnormalities: Clues to Molecular Pathogenesis?

To the Editor:

In reference to the paper by Elliott et al. [1996], we believe that some cases of circumferential ringed creases (or "Michelin tire babies") with specific histologic findings and/or karyotype abnormalities may provide clues as to the molecular pathogenesis in at least some of these patients. In a karyotypically abnormal child with multiple anomalies, mental retardation, hypertrichosis, and circumferential ringed creases, we found an underlying smooth muscle hamartoma in a skin biopsy of one of the creases [Schnur et al., 1993—also reported as case 2 of Cohen et al., 1993]. This same histologic pattern was documented in 4 other children with circumferential skin creases in reports by Wallach et al. [1980; Glover et al. [1989], Patrizi et al [1989], and Oku et al. [1993]. Of these five cases, 2 patients beside ours [reports by Wallach et al., 1980; Oku et al., 1993] had mental retardation and/or seizures, as well as other anomalies and in two patients [reports by Glover et al., 1989; Patrizi et al., 1989], the phenotype was limited to the skin.

An even more recent paper in this journal by Pivnick et al. [1996] reports a cytogenetically normal, developmentally delayed child whose photographs clearly show ringed creases of the extremities in addition to his other anomalies. Although the creases were not specifically discussed by the authors, a skin biopsy demonstrated a smooth muscle hamartoma; the observed hypertrichosis is also consistent with this. Thus, this child should also be considered as being similar to others with the "Michelin tire" type of circumferential creases.

In the group of patients who have smooth muscle hamartomas underlying their skin creases, a contiguous gene syndrome hypothesis could be invoked to explain the variable spectrum of clinical findings. Although no readily apparent deletions were detected in the patient reported in Schnur et al. [1993], a clue to localization of a gene which causes these hamartomatous skin creases may have been provided by the karyotype of that child, a seemingly balanced, maternally transmitted paracentric inversion of chromosome 7. Alternatively, this finding may have been coincidental to the phenotype.

We strongly suggest that both karyotypes and skin biopsies be performed on all children with ringed skin creases, with and without other anomalies, to attempt to delineate the molecular etiology of this disorder or group of disorders.

REFERENCES

Cohen MM, Gorlin RJ, Clark R, Ewing SG, Camfield PR (1993): Multiple circumferential skin folds and other anomalies: A problem in syndrome delineation. Clin Dysmorphol 2:39–46.

Elliott AM, Ludman M, Teebi AS (1996): New syndrome?: MCA/MR syndrome with multiple circumferential skin creases. Am J Med Genet 62:23–25.

Glover MT, Malone M, Atherton DJ (1989): Michelin-tire baby syndrome resulting from diffuse smooth muscle hamartoma. Pediatr Dermatol 6:329–331.

Oku T, Iwasaki K, Fujita H (1993): Folded skin with an underlying cutaneous smooth muscle hamartoma. Brit J Dermatol 129:606–608.

Patrizi A, Neri I, Varotti C (1989): Un autre case de naevus musculaire généralisé avec aspect clinique de "bébé Michelin" associé a une mastocytose cutanée. Ann Dermatol Vénéréol 116:551–554.

Pivnick EK, Wilroy RS, Martens PR, Teather TC, Hashimoto K (1996): Hypertrichosis, pigmentary retinopathy, and facial anomalies: A new syndrome? Am J Med Genet 62:386–390.

Schnur RE, Herzberg AJ, Spinner H, Kant JA, Magnusson M, McDonald-McGinn D, Rehberg K, Honig PJ, Zackai EH (1993): Variability in the Michelin tire syndrome. J Am Acad Dermatol 28:364–370.

Wallach D, Sorin M, Saurat JH (1980): Naevus musculaire généralisé avec aspect clinique de "bébé Michelin." Arch Dermatol Vénéréol (Paris) 107:923–927.

Rhonda E. Schnur
Elaine H. Zackai
Department of Pediatrics and Genetics
The Children's Hospital of Philadelphia and
The University of Pennsylvania School of Medicine
Philadelphia, Pennsylvania

*Correspondence to: Rhonda E. Schnur, Division of Genetics, Cooper Hospital/University Medical Center, 636-638 Benson St., Camden, NJ 08103.

Received 3 June 1996